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Case Report

Prenatal diagnosis of bilateral anophthalmia by 3D “reverse face” view ultrasound and magnetic resonance imaging

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Abstract

Objective: Primary anophthalmia is a rare congenital malformation that affects 0.6/10,000 liveborn infants. It is usually associated with central nervous system malformations, aneuploidies, cytomegalovirus infection and mental retardation and it can also be part of genetic conditions such as Fraser, Goltz, Goldenhar, Waardenburg and Lenz syndromes. Neonatal prognosis depends on whether anophthalmia is an isolated malformation, or it is associated with other defects or part of a syndrome.

Case Report: A healthy 43-year-old woman, G4 P3 with three previous healthy children, was referred to our clinic for a routine obstetric ultrasound at 28 weeks' gestation. The fetal eye globes and lenses could not be seen on two-dimensional (2D) ultrasound, which led to the diagnosis of bilateral congenital anophthalmia. No other fetal malformations were detected. At 30 weeks' gestation, a three-dimensional (3D) ultrasound was performed using the rendering mode and “reverse face” view. Using this technique, the absence of both eye globes could be clearly seen through a “slit”. 3D-ultrasound allowed the parents to better understand their child's problem and possible postnatal implications. Fetal magnetic resonance imaging (MRI) was also performed, to study the fetal cortex in more detail. This exam revealed right cerebral hemisphere sulci and gyri hypoplasia. At 41 1/7 weeks, she went into spontaneous labor and delivered vaginally a 3525 g male infant with Apgar scores of 9 and 10. Postnatal exams confirmed bilateral congenital anophthalmia.

Conclusion: This is the first case report in the literature of prenatal diagnosis of bilateral anophthalmia using 3D “reverse face” view ultrasound and MRI.

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Keywords: anophthalmia; magnetic resonance imaging; prenatal diagnosis; three-dimensional ultrasound

Introduction

Ultrasound has been a very important tool in the prenatal diagnosis of malformations, especially after the improvement in the quality of the images. It is part of the fetal sonographic examination, with visualization of the orbits and lenses of fetuses; then it is possible to diagnose many defects such as cyclopia, microphthalmia, cataracts, and anophthalmia [1,2].

Anophthalmia diagnosis can be made by two-dimensional (2D) ultrasonography when eyeballs and lens are absent, but using three-dimensional (3D) ultrasonography we have more information, especially when the fetal head position is unfavorable [3,4]. The use of 3D-ultrasound is well known for prenatal diagnosis of fetal facial abnormalities [3]. The technique of 3D “reverse face” view was firstly described by Campbell et al [5] for the diagnosis of cleft palate, but it can also be used for visualization and evaluation of the orbits of the fetus; it has the advantage of not being affected by the problem of shadowing [4]. It is necessary to take a satisfactory 3D volume with adequate inclusion of both orbits [3].

Fetal magnetic resonance imaging (MRI) is a tool that can be also used, especially in cases of malformations of the

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central nervous system (CNS), bringing new information about the abnormalities found and detailed evaluation of brain sulci and gyri. MRI confirms the absence of eye tissue, the optic nerve and extraocular muscles in cases of anophthalmia [6].

Case report

A 43-year-old healthy woman, gravida 4 para 3, was referred to our service (São Paulo Hospital, São Paulo Federal University - UNIFESP, São Paulo, SP, Brazil) for her first obstetrics ultrasound scan at 28 weeks' gestation. When the eyeballs and lenses were not seen, bilateral anophthalmia was diagnosed. No other fetal malformations were found. The scan was performed in the division of fetal neurology of the department of obstetrics, by an examiner (EAJ). The Voluson 730 Expert (General Electric Medical System, Zipf, Austria) machine, equipped with a convex volumetry transducer (RAB 4-8L), produced the images in 2D- and 3D-ultrasound, using the 3D "reverse face" technique (Figs. 1 and 2). This permitted better visualization of the bilateral anophthalmia. The fetal MRI was produced in the department of diagnostic imaging, using a 1.5 T Sonata Maestro Class (Siemens Medical Solutions, Erlangen, Germany) by an examiner (PSO), without previous knowledge of the results of the ultrasound. The MRI of the fetal brain showed underdevelopment of the sulci and gyri of the right hemisphere, in addition to bilateral anophthalmia (Fig. 3).

Fetal karyotype was not performed, because the patient could not afford the examination. The patient denied similar cases in the family, drug addiction, medication, or contact with chemicals during pregnancy. She reported that she only smoked early on in the pregnancy and she has an autistic son.

The pregnant woman underwent vaginal delivery at 41 weeks; a male, 3525 g, Apgar = 9–10, was born (Fig. 4). In

the postnatal investigation, a computerized tomography scan confirmed bilateral anophthalmia and cortical dysplasia. Echocardiography also diagnosed bicuspid aortic valve.

Infants are referred for follow-up with neuropsychiatrics/neurosurgery, ophthalmology (to evaluate ocular prosthesis during growth and development of the child) and genetics.

Discussion

Isolated anophthalmia is a rare event with an incidence of 0.6/10,000 live births [7]. It can also be found in association with other malformations, and a syndrome can be characterized, such as Fraser [8], Goltz, Goldenhar, Waardenburg and Lenz syndrome, defining the prognosis of the affected child. There is often a family history and it is possible to define autosomal dominant or recessive inheritance in these cases [9].

The 3D image can be seen in multiplanar and rendering modes. 3D "reverse face" view was described firstly by Campbell et al [5], to assess fetuses with cleft palate. This technique consists of obtaining a sagittal view of the fetal face, with the green line (ROI – region of interest) put through the alveolar ridge and palatal area. This image is then rotated 180° in the "z" axis, obtaining the 3D "reverse face" view. This technique was used by Wong et al [4] in two cases of unilateral anophthalmia; it proved to be invaluable in providing a quick and easy diagnosis in a case where the fetal head position was unsatisfactory. Since the 3D "reverse face" view is relatively free of the problem of shadowing [5], a satisfactory 3D volume taken at the acquisition, with adequate inclusion of both orbits, will enable an effective examination of the orbits and eyeballs. In our case, the 3D "reverse face" view was obtained quickly, demonstrating bilateral anophthalmia due to the absence of both orbits. This is the first case in literature of the prenatal diagnosis of bilateral anophthalmia using the 3D "reverse face" view.



Fig. 1. Fetus with bilateral anophthalmia by ultrasound. To the right, the coronal image of the two-dimensional ultrasound of the face of the fetus and to the left, the 3D-dimensional rendering the image of the face of the fetus.



Fig. 2. Fetus with bilateral anophthalmia by 3D “reverse face” view technique. To the right, the sagittal plane of face of the fetus is rotated 180° in the “z” axis, and the green line (ROI — region of interest) is placed at the level of fetal orbits. To the left, the resulting image of bilateral anophthalmia by 3D “reverse face” view.

The MRI proved by T2 [spin echo-based half-Fourier acquisition single-shot turbo spin-echo (HASTE) and gradient echo-based steady-state free precession (true FISP)] weighted sequences obtained in axial, sagittal and coronal planes the bilateral anophthalmia. The MRI demonstrated unilateral

anophthalmia in a case of otocephaly in a fetus with 29 weeks of gestation. The MRI enabled a precise assessment of fetal facial and ocular abnormalities (microphthalmia/anophthalmia) [10]. This is the first case in the literature of the prenatal diagnosis of bilateral anophthalmia using MRI.

In summary, we present a first case in the literature of the prenatal diagnosis of bilateral anophthalmia using 3D “reverse face” view and MRI. The initial prenatal diagnosis of anophthalmia should be done by 2D ultrasound, as no discrete globe or lens is detectable; the 3D method or MRI should be used as adjuvant techniques to confirm the diagnosis. For parents, the prenatal diagnosis of anophthalmia, as well as other malformations, is very important, as this significantly influences the acceptance of the child and psychological preparation for his arrival. In the case of anophthalmia, ocular prosthesis is often necessary for the normal development of bones in the skull, avoiding deformities during growth. Thus, a precise prenatal diagnosis is important for guiding parents regarding

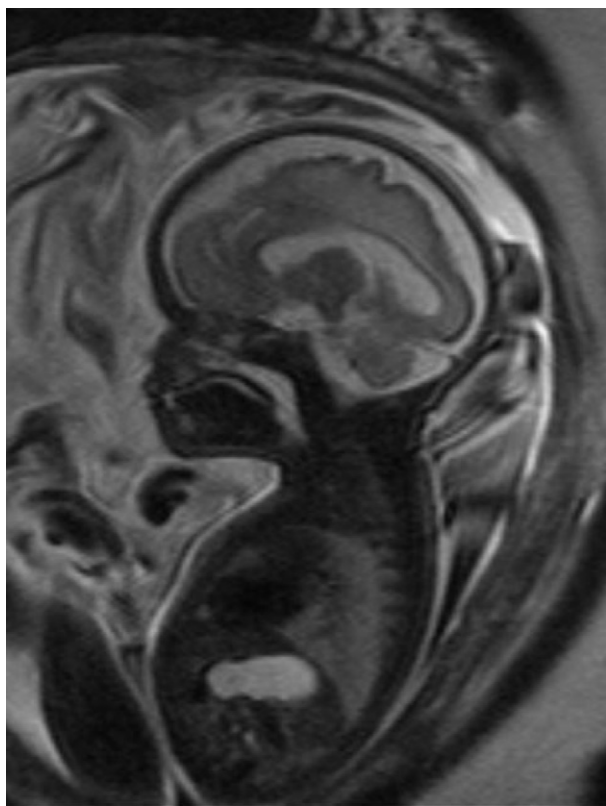


Fig. 3. Fetus with bilateral anophthalmia by MRI. T2 [spin echo-based half-Fourier acquisition single-shot turbo spin-echo (HASTE) and gradient echo-based steady-state free precession (true FISP)] weighted sequence in sagittal plane, proving the bilateral anophthalmia.



Fig. 4. Newborn with bilateral anophthalmia.

the prognosis and management of the child and for genetic counseling for future pregnancies.

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